

I claim:

1. A method of detecting mutant nucleic acid comprising:

a) contacting a sample comprising nucleic acid with mutant PCR primers,

b. amplifying the product of step a) under short PCR conditions,

and

c. identifying the presence of amplicons of step b), wherein the presence of such amplicons is indicative of the presence of nucleic acid deletion sequences.

2. The method of claim 1 wherein the nucleic acid is mitochondrial nucleic acid.

3. The method of claim 2 wherein the nucleic acid is mt-DNA.

4. The method of claim 1 further comprising the step of contacting the sample with a cleavage reagent.

5. A method of detecting nucleic acid having a deletion comprising:

a) obtaining a sample comprising nucleic acid;

b) dividing the sample into a first aliquot and a second aliquot, each suspected of containing a mixture of mutant DNA and wild type DNA;

c) contacting the first aliquot with a cleavage reagent, thereby forming a mixture;

d) contacting the mixture of step c) with a forward primer complementary to a priming site upstream of the deletion sequence;

- e) to the mixture of step d), adding a reverse primer complementary to the downstream zone of the mutant DNA and each of four different nucleoside triphosphates as well as a DNA polymerase, under conditions such that only the mutant DNA is amplified;
- f) contacting the second aliquot with a forward primer complementary to a priming site upstream of the deletion sequence and a reverse primer complementary to a priming site within the deletion sequence;
- g) to the mixture of step f) adding four different nucleoside triphosphates, and a DNA polymerase under conditions such that the DNA is amplified; and
- h) detecting the presence of the amplified DNA.

6. The method of claim 5 further comprising the steps of contacting each aliquot with probes and detecting the presence or absence of the probes.

7. The method of claim 5 wherein detection is conducted by gel electrophoresis or capillary electrophoresis.

8. The method of claim 6 wherein the probes comprise a member of the group consisting of TaqMan probes, molecular beacons, PNA probes, DNAzymes, and combinations thereof.

9. A method of detecting nucleic acid having a deletion comprising:

- a) obtaining a sample comprising nucleic acid;
- b) dividing the sample into a first aliquot and a second aliquot, each suspected of containing a mixture of mutant DNA and wild type DNA;
- c) contacting the first aliquot with a cleavage reagent, thereby forming a mixture;
- d) contacting the mixture of step c) with a reverse primer downstream of the deletion sequence;
- e) to the mixture of step d), adding a forward primer complementary to the region upstream of the deletion sequence and each of four different nucleoside triphosphates as well as a DNA polymerase, under conditions such that only the mutant DNA is amplified;
- f) contacting the second aliquot with a forward primer complementary to a priming site within the deletion sequence, and a reverse primer downstream of the deletion sequence;

g) to the mixture of step f) adding four different nucleoside triphosphates, and a DNA polymerase under conditions such that the DNA is amplified; and
g) detecting the presence of the amplified DNA.

5 10. The method of claim 9 further comprising the steps of contacting each aliquot with probes and detecting the presence or absence of the probes.

11. The method of claim 9 wherein detection is conducted by gel electrophoresis or capillary electrophoresis.

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12. The method of claim 10 wherein the probes comprise a member of the group consisting of TaqMan probes, molecular beacons, PNA probes, DNAzymes, and combinations thereof.

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13. A method of quantitating nucleic acids having deletion sequences comprising:

a) contacting an aliquot of sample comprising nucleic acid with mutant PCR primers under short PCR conditions,

b) amplifying the product of step a),

c) contacting a different aliquot of said nucleic acid sample comprising nucleic acid with wild type PCR primers,

d) amplifying the product of step c),

25 e) identifying the presence of amplicons of step b), and

f) quantitating the presence of amplicons of step b).

14. The method of claim 13 further comprising the step of contacting the aliquot of step a) or step
30 b) with a cleavage reagent.

15. The method of claim 13 further comprising the step of identifying the presence of amplicons of step d), and wherein the quantitation of the presence of amplicons of step b) is relative to the amount of wt nucleic acid present in the sample.

5 16. The method of claim 13 wherein quantitation is conducted by comparison to a standard.

17. The method of claim 13 wherein quantitation is conducted by real-time monitoring.

10 18. A composition of matter selected from the group consisting of Seq. ID 2, Seq. ID 3, Seq. ID 4, Seq. ID 6, Seq. ID 7, Seq. ID 8, Seq. ID 9, and Seq. ID 10, Seq. ID 11, Seq. ID 12, Seq. ID 13, Seq. ID 15, Seq. ID 16, Seq. ID 17, Seq. ID 19, Seq. ID 21, Seq. ID 22, Seq. ID 23, Seq. ID 24, Seq. ID 25, Seq. ID 27, Seq. ID 28, Seq. ID 29, Seq. ID 30, Seq. ID 31 and Seq. ID 32.

15 19. A kit for detecting or quantitating the occurrence of nucleic acid deletion sequences comprising mutant PCR primers.

20 20. The kit of claim 19 further comprising short PCR reagents.

25 21. The kit of claim 19 comprising a composition selected from the group consisting Seq. ID 2, Seq. ID 3, Seq. ID 4, Seq. ID 6, Seq. ID 7, Seq. ID 8, Seq. ID 9, and Seq. ID 10, Seq. ID 11, Seq. ID 12, Seq. ID 13, Seq. ID 15, Seq. ID 16, Seq. ID 17, Seq. ID 19, Seq. ID 21, Seq. ID 22, Seq. ID 23, Seq. ID 24, Seq. ID 25, Seq. ID 27, Seq. ID 28, Seq. ID 29, Seq. ID 30, Seq. ID 31 and Seq. ID 32.